

Hypermethioninemia (MET)

An amino acid disorder

What is it?

Hypermethioninemia (also known as MET) is an inherited amino acid disorder. People with amino acid disorders, like MET, cannot properly break down certain components of protein. This is because the body is lacking a specific chemical (enzyme). Since the body cannot properly break down the protein, certain amino acids build up in the blood and urine and cause problems when a person eats normal amounts of protein.

What are the symptoms?

People with MET usually have no symptoms. There have been reports of people with MET having foul breath, and having problems with the insulation surrounding the brain. People with MET typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

Inheritance and frequency

MET is inherited in an autosomal recessive manner. This means that for a person to be affected with MET, he or she must have inherited two non-working copies of the gene responsible for causing MET. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have MET. Typically, there is no family history of MET in an affected person. The number of people affected with MET is unknown.

How is it detected?

MET may be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

How is it treated?

MET may be treated by eating a diet low in protein and they may be given a special formula, as recommended by a genetic metabolic medical professional.

DISCLAIMER: This information is not intended to replace the advice of a genetic metabolic medical professional.

For more information:

Genetics Home Reference

Website: <http://ghr.nlm.nih.gov/ghr/page/Home>

Save Babies Through Screening Foundation

4 Manor View Circle Malvern, PA 19355-1622 Toll Free Phone: 1-888-454-3383 Fax:

(610) 993-0545 Email: email@savebabies.org

Website: <http://www.savebabies.org/diseasedescriptions.php>

American College of Medical Genetics

Newborn Screening ACT Sheets and Confirmatory Algorithms

<http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm>